

Appendix S2. Phenotypic comparison of mutations found in our cohort with the literature.

	MPZ G163R		MPZ R98H		MPZ D224Y		LITAF V144M		GJB1 R183S		NEFL Q332P		INF2 L77R		MFN2 R280H		MFN2 R94Q	
	CS	Lit	CS	Lit	CS	Lit	CS	Lit	CS	Lit	CS	Lit	CS	Lit	CS	Lit	CS	Lit
CMT type	1	1 [1,2]	1, 2	1 [3]	1	1* [4]	1	1 [5]	1(M)	2(F) [6]	1	2 [7]	DI	1 [8]	2	2 [9,10]	2	2 [9,11]
Age of onset (y)	39, childhood	1st-7th decade [1,2]	20-25, 62	early-onset -60 [3]	40	AAE 31,78 [4]	29,49	10-57 [5]	childhood	na	26	2nd-3rd decade [7]	AAE 12	4 [8]	teens	11-35 [9,10],	5	2-17 [9,11]
Hypertrophic nerve roots	+	-	+	-	-	-	-	-	na	-	-	-	-	-	-	-	-	
CSF protein elevation	+	-	+	[12]	-	[4]	+	-	na	-	-	-	-	-	-	-	-	
Cataracts bilateral	+	-	+	-	-	-	-	-	na	-	-	-	-	-	-	-	-	
CTS	+	[1]	-	-	-	-	+	-	na	-	-	-	-	-	-	-	-	
Pain	-	-	-	[12]	-	-	+	[5]	+	na	-	-	-	-	+	[10]	-	
Paresthesias	-	[1,2]	+	-	-	-	+	[5]	+	na	+	-	-	-	+	-	-	
Tremor	+	-	-	-	-	[4]	+	-	+	na	-	-	-	-	-	[10]	+	[9]
Respiratory insufficiency	-	-	+	-	-	-	-	-	na	-	-	-	-	-	-	-	-	
Scoliosis	+	-	-	-	-	-	+	-	na	-	-	-	-	-	-	-	-	
Hip dysplasia	-	-	-	[13]	-	-	-	-	-	-	-	-	-	-	-	-	-	
Fasciculations	-	-	+	-	-	-	+	-	na	-	[7]	-	-	-	-	-	-	
FSGS	-	-	-	-	-	-	-	-	na	-	-	+	[8]	-	-	-	-	
Pupillary abnormalities	+	-	-	-	-	-	-	-	na	-	-	-	-	-	-	-	[14]	
RLS	-	-	-	-	-	-	+	-	na	-	-	-	-	+	-	-	-	
BrainMRI abnormalities	-	-	+	-	-	-	-	-	na	-	-	-	[8]	-	[10]	-	-	
Claw hands	-	-	+	-	-	-	-	-	na	-	-	-	-	-	-	-	-	
Asymmetrical weakness	-	-	-	[12]	-	-	-	-	na	-	-	-	-	-	-	-	-	
Autonomous	-	-	-	-	+	-	+	-	na	-	-	-	-	-	-	-	+	
Severe slow NCV	-	-	-	-	-	-	-	-	na	-	-	-	-	-	-	-	-	
UL predominant	-	-	-	-	-	-	+	-	na	-	-	-	-	-	-	-	-	
Facial weakness	-	-	-	-	-	-	-	-	na	-	[7]	-	-	-	-	-	-	
Deafness	-	-	-	-	-	-	-	-	na	-	-	-	[8]	-	[10]	-	-	
Hyperkeratosis	-	-	-	-	-	-	-	-	na	-	[7]	-	-	-	-	-	-	
Syndactyly	-	-	-	-	-	-	-	-	na	-	[7]	-	-	-	-	-	-	
Cold induced hand cramps	-	-	-	-	-	-	-	-	na	-	-	-	-	+	-	-	-	
Bulbar	-	-	-	-	-	-	-	-	na	-	-	-	-	-	[10]	-	-	
Upper motor neuron	-	-	-	-	-	-	-	-	na	-	-	-	-	-	[10]	-	-	
Migraine	-	-	-	-	-	-	-	-	na	-	-	-	-	-	[10]	-	-	
Cognitive impairment	-	-	-	-	-	-	+	-	na	-	-	-	-	[8]	-	-	-	
MS as additional diagnosis	-	-	-	-	-	[4]*	+	-	na	-	-	-	-	-	-	-	-	

CS, current study; Lit, literature; CMT, Charcot-Marie-Tooth neuropathy; *homozygous patient(s); M, male; F, female; na, not available; DI, dominant intermediate; AAE, age at examination; CSF, cerebrospinal fluid; CTS, carpal tunnel syndrome; FSGS, focal segmental glomerular sclerosis; RLS, restless legs syndrome; MRI, magnetic resonance imaging; NCV, nerve conduction velocity; UL, upper limb; MS, multiple sclerosis. For abbreviations of genes see text.

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